Digitalising Genetic Counselling

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Abstract book

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Contents

iSCREEN: Digital screening for familial cancer may empower patients, unburden health professionals, and improve preventive efforts

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Background: With a third of breast cancer patients presenting with a familial cancer burden, early identification of familial risk is crucial for effective prevention. In Germany, physicians currently use the German Cancer Society's checklist to take a familial cancer history. Digitizing this checklist and actively involving patients could streamline preventive efforts.

Objective: We compare the accuracy of iSCREEN, a digital, patient-facing implementation of the German Cancer Society's checklist, to physician-led interviews based on the checklist. We also assess iSCREEN's usability and user experience (UUX).

Methods: We transformed the checklist for physicians into a digital, chatbot-like questionnaire for patients. We first tested its accuracy with 200 pedigrees in-silico. Second, we asked 160 patients to use iSCREEN and report on its UUX and compared accuracy against physician-led interviews.

Results: iSCREEN identified all patients with familial cancer risk in both studies. Patients found the tool comprehensible, easy to use, and reported a preference for its future utilization. No adverse psychological effects were recorded.

Conclusion: iSCREEN presents an accurate, easy-to-use digital solution for the identification of familial cancer risks. Adoption of iSCREEN may empower patients, unburden health professionals, and ultimately improve prevention of familial cancer thanks to early identification of persons at risk.

Evaluation of the health-related DTC-GT market & information provision for Dutch consumers: (un)informed decisions?

Authors: Bruins, D.[1], Onstwedder, S.[1,2], Cornel, M.[1], Ausems, M.[3], Van Mil, M.[4,5], Rigter, T.[1,2]

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Background & Methods: Health-related direct-to-consumer genetic testing (DTC-GT) is surrounded by ample risks and opportunities for individuals, society, and the public health care system. Combined with a lack of regulative and legislative unison regarding DTC-GT between countries worldwide and high market dynamicity, it is paramount that potential consumers can make an informed decision about consuming such tests. To this end, we aimed to evaluate the current health-related DTC-GT market for Dutch consumers, and subjected the websites of several large players identified during this market analysis to an in-depth content analysis to assess information provision.

Preliminary results: We found that there are currently approximately 10 large players in the health-related DTC-GT market for Dutch consumers. The websites of three sellers are currently being subjected to content analysis. The overall provision and presentation of information appears to differ noticeably between sellers, indicating potential for improvement to better enable potential Dutch consumers to make informed decisions regarding DTC-GT usage. This would allow to better mitigate the potential risks of DTC-GT for individuals and society. We aim to publish our final findings, suggestions, and conclusions in the Special Issue 'Human Genetics: Diseases, Community, and Counseling' of the journal 'Genes' at the end of 2023.

Design and evaluation of a virtual genetic counsellor

Authors: Freriks, S.[1]

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Abstract:

In this master's thesis research, the central question revolves around the concept of "embodiment" within the context of a virtual genetic counsellor. To begin, it is essential to clearly understand what "embodiment" means in this specific context. This foundational step lays the groundwork for the subsequent research.

To gain insights into the preferences and expectations of potential users, focus group discussions were conducted. These discussions served as a platform for individuals to share their perspectives and experiences related to genetic counselling.

A thematic analysis was undertaken following the data collection from the focus group discussions. This analytical process aimed to uncover the fundamental and implicit needs and desires of individuals interacting with a virtual genetic counsellor.

The research progressed to a practical phase. An interactive prototype of the virtual genetic counsellor was developed. This prototype represented the envisioned counsellor, incorporating the insights gained from the focus groups and thematic analysis.

To evaluate the prototype, task-based evaluations were conducted. These sessions provided valuable feedback and shed light on areas that required refinement.

Finally, the research culminated in a comprehensive analysis of the evaluation results, to draw conclusions about the ideal embodiment of a virtual genetic counsellor.

dVP_FAM: A digital platform to improve the care pathway of patients with familial cancer risks in Germany

Authors: Mangum, C.[1,2], Amirpour-Mehrhof, N.[2], Klein, K.[3,6], Thomas, P.[3], Stegen, S.[2,5], Wagner, J.K.[2], Blohmer, J.U.[2], Kendel, F.[3], Speiser, D.[2], Feufel, M.A.[1]

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Abstract:

In the evolving landscape of genetic medicine, continuous, seamless, and individualized care for patients with familiar cancer risks is paramount to improve risk management and ideally prevent cancer. To circumvent limitations of the current, mainly analogue care pathways, we developed dVP_FAM, a cutting-edge digital platform tailored to streamline the identification and guidance of individuals facing familial cancer risks throughout their entire care pathway in the German health care system. As a starting point, an easy-to-use digital screening tool (iSCREEN) supports community gynecologists to identify patients with family cancer burden. Once identified, patients at risk may use the platform to manage and share documents and to make appointments at specialized counseling centers. Users maintain control over their data and have access to pertinent, personalized information throughout their entire care pathway from local clinics to specialized counseling centers and back. Based on an outline of the platform features, we describe the cluster-randomized mixed methods study starting in October 2023 to assess the effectiveness and effectivity of the platform and its impact on patient empowerment and user experience. After positive evaluation, the dVP_FAM may be transferred to standard care and serve as a blueprint for digitizing care pathways of other genetic cancers.

Be Prepared – The effect of prior animations on genetic counseling in at-risk women and men, who are offered pre-symptomatic BRCA1 or BRCA2 testing: a study protocol

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[1] Department of Clinical Genetics, Erasmus MC Cancer Institute, University Medical Center Rotterdam

Abstract:

Rationale
Strategies to improve the efficiency of genetic counseling are needed because of long waiting lists for clinical genetic centers. Patients who are referred for pre-symptomatic testing (BRCA1/2) for breast and ovarian cancer are at risk up to 50% to be carrier of the familial mutation. The information given during genetic counseling can be complex. Using animations as a preparatory stage for genetic counseling with information can possibly help to make an informed decision.

Objective
The objective of this study is to evaluate whether offering animations with information about a BRCA1/BRCA2 mutation before genetic counseling improves empowerment, satisfaction and efficiency.

Study type
In a randomized controlled trial, an initial N = 144 patients will be assigned to the intervention group or control group. The control group receives conventional pre-symptomatic counseling, while the intervention group will be offered animations with information about BRCA1/BRCA2 before counseling.

Study population
Patients referred to the department of Clinical genetics of the Erasmus Medical Centre for DNA-testing for familial BRCA1/BRCA2 mutation.

Methods
Participants are invited to complete an online questionnaire before genetic counseling and after genetic testing. The primarily outcome measure is the effect on patient empowerment measured with the GCOS-18, patient satisfaction and efficiency.

Informing family members at risk: the right information by the right person at the right time

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Abstract:

An index patient currently receives a paper letter for family members at-risk, with information about the condition, treatment options and the possibility for DNA testing. Family members can take this letter to their GP for a referral to a clinical genetics department. However, only half of family members at risk are actually being referred for genetic counseling. Reasons for non-referral may include the fact that the right information doesn't reach the family members or isn't understood by them.

To improve this information process, a digital family letter is being developed which index patients can email to their relatives. Extra attention is being paid to low literacy. The text is written at B1 language level. Furthermore, the digital layered approach makes it possible to clearly show the most important information. Background information is available by clicking on certain topics. The digital letter also includes a link to an animation in which the care path is visualized.

The digital family letter is initially being developed for Lynch syndrome and implemented in the clinical genetics department of Radboud university medical center. After evaluation and if needed optimalisation, the digital letter can be developed for other conditions and implemented in other clinical genetics departments.

Supporting non-genetic healthcare professionals in mainstreaming of breast cancer

Authors: Vlaming, M.[1], Bokkers, K.[1], Bos, M.[1], Fransen van de Putte, D.[1], Ausems, M.[1]

[1] Department of Genetics, University Medical Center Utrecht

Abstract:

The number of germline genetic tests is increasing. To meet the needs of these growing numbers, new genetic testing pathways are being developed. Mainstream genetic testing is one of them. In a mainstream genetic testing pathway, genetic testing is not discussed and requested by a clinical geneticist, but by a non-genetic healthcare professional. To support healthcare professionals in mainstreaming, UMC Utrecht has launched a website (dna-mainstream.umcutrecht.nl). Here, healthcare professionals can get background information and information on how to provide pre-test genetic counseling or when to refer a patient to the genetics department. This website is implemented in genetic care pathways for patients with breast or ovarian cancer, and can be adapted for other cancer types like prostate cancer. In addition, the website contains an online genetic testing request form for breast cancer patients. After answering several questions on the criteria for genetic testing, healthcare professionals are informed whether a patient is eligible for genetic testing. If so, a PDF file is ready to be downloaded to send to the genetics department and use as a referral form.

Designing an intuitive dialogue management interface for AI-collaborative content generation in eHealth conversational agents

Authors: Zhao, X.[1]

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Abstract:

This research presents the design and evaluation of an intuitive dialogue management interface integrated with a Pre-trained Language Model (PLM) for non-technical medical experts. The interface facilitates the effective crafting of dialogues for eHealth conversational agents, focusing on the eCG Family Clinic project as a case study. The graphical user interface (GUI) and text editor components provide a seamless and synchronized interaction experience, enabling users to manage and edit dialogue content effortlessly. Prompt engineering techniques are employed to structure user requests using prompt templates, leveraging the capabilities of the PLM. Connectivity between the interface and the WOOL platform enables the conversion of dialogue diagrams into scripts suitable for conversational agents. The evaluation, based on feedback from medical experts, identifies areas for improvement such as scalability of the GUI, prompt management usability, and content quality generated by the PLM. In light of these valuable insights, the dialogue management interface demonstrates its potential as a valuable tool for non-technical medical experts, empowering them to design informative and inclusive eHealth conversational agents.

Additional posters

Designing a virtual assistant for digital genetic counseling

Beinema, T.[1], Klaassen, R.[1], Kolkmeier, J.[1], Van Lingen, M.[2], Van den Heuvel, L.[2], Siemelink, M.[2], Van Essen, D.[3], Van Tintelen[2], P., Heylen, D.[1]

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Digital technologies in genetic counselling: an overview of ethical aspects

Van Lingen, M.[1], Giesbertz, N.[2], Bredenoord, A.[3], Van den Heuvel, L.[1], Van Tintelen, P.[1], Jongsma, K.[4]

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DNA-poli: an online platform for families at risk of inheritable cardiac diseases

Van Lingen, M.[1], Siemelink, M.[1], Van den Heuvel, L.[1], Giesbertz, N.[1], Van Tintelen, P.[1], Beinema, T.[2], Kolkmeier, J.[2], Klaassen, R.[2], Heylen, D.[2]

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